# THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL[1] DG-5.0.0 (57 GENES)

| Gene | Twist X2 covered 10x | Twist X2 covered 20x | srWGS covered 10x | srWGS covered 15x | srWGS covered 20x | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|---|---|---|
| ABL1 | 100% | 100% | 100% | 100% | 99.7% | Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602 |
| ACTA2 | 100% | 100% | 100% | 100% | 99.8% | Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042 |
| ARIH1 | 100% | 100% | 100% | 100% | 99.7% | |
| ASPH | 100% | 100% | 100% | 100% | 99.7% | Traboulsi syndrome, 601552 |
| BGN | 99.9% | 98.5% | 97.4% | 85.2% | 66.2% | Meester-Loeys syndrome, 300989;Spondyloepimetaphyseal dysplasia, X-linked, 300106 |
| CBS | 88% | 85.8% | 100% | 99.9% | 99.2% | Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200 |

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| COL1A1 | 100% | 100% | 100% | 100% | 99.1% | Osteogenesis imperfecta, type II, 166210;Caffey disease, 114000;Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;{Bone mineral density variation QTL, osteoporosis}, 166710;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type III, 259420 |
| COL1A2 | 100% | 100% | 100% | 100% | 99.7% | Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210 |
| COL3A1 | 100% | 100% | 100% | 99.9% | 99.7% | Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343 |

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| COL5A1 | 100% | 100% | 100% | 100% | 99.3% | Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329 |
| COL5A2 | 100% | 100% | 100% | 100% | 99.8% | Ehlers-Danlos syndrome, classic type, 2, 130010 |
| DLG4 | 100% | 100% | 100% | 100% | 99.2% | Intellectual developmental disorder, autosomal dominant 62, 618793 |
| EFEMP1 | 84.9% | 84.9% | 100% | 100% | 99.9% | Doyne honeycomb degeneration of retina, 126600;Cutis laxa, autosomal recessive, type ID, 620780;Glaucoma 1, open angle, H, 611276 |
| EFEMP2 | 99.9% | 97.3% | 100% | 99.9% | 99.2% | Cutis laxa, autosomal recessive, type IB, 614437 |
| ELN | 100% | 99.9% | 100% | 99.9% | 99.4% | Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500 |
| EMILIN1 | 100% | 100% | 100% | 99.8% | 99% | Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080;Arterial tortuosity-bone fragility syndrome, 620908 |

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| FBN1 | 100% | 100% | 100% | 100% | 99.8% | Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900 |
| FBN2 | 99.2% | 99.2% | 100% | 100% | 99.7% | Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050 |
| FKBP14 | 100% | 100% | 100% | 100% | 100% | Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 |
| FLNA | 100% | 100% | 98.4% | 87.4% | 67% | Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620 |

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| FOXE3 | 100% | 100% | 100% | 99.5% | 95.8% | Anterior segment dysgenesis 2, multiple subtypes, 610256;{Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349;Cataract 34, multiple types, 612968 |
| FURIN | 100% | 99.7% | 100% | 100% | 99.5% | |
| HCN4 | 100% | 100% | 100% | 100% | 98.8% | Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123 |
| IPO8 | 97% | 97% | 100% | 100% | 99.7% | VISS syndrome, 619472 |
| JAG1 | 100% | 100% | 100% | 100% | 99.7% | ?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500 |
| KANSL1 | 98.1% | 98.1% | 100% | 100% | 99.8% | Koolen-De Vries syndrome, 610443 |
| LMOD1 | 100% | 100% | 100% | 100% | 98.9% | ?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362 |
| LOX | 100% | 100% | 100% | 99.9% | 99.1% | Aortic aneurysm, familial thoracic 10, 617168 |
| LTBP3 | 100% | 100% | 100% | 99.9% | 98.8% | Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809 |
| MAT2A | 100% | 100% | 100% | 100% | 99.9% | |

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| MED12 | 100% | 100% | 98.3% | 86.8% | 66.7% | Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450 |
| MFAP5 | 100% | 100% | 100% | 100% | 99.9% | Aortic aneurysm, familial thoracic 9, 616166 |
| MYH11 | 100% | 100% | 100% | 100% | 99.4% | Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350 |
| MYLK | 99.2% | 99.2% | 100% | 99.9% | 99.5% | Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210;Aortic aneurysm, familial thoracic 7, 613780 |
| NOTCH1 | 99.1% | 99% | 100% | 100% | 99.2% | Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730 |
| NPR2 | 100% | 100% | 100% | 100% | 99.6% | Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875 |
| NPR3 | 100% | 100% | 100% | 99.8% | 98.5% | Boudin-Mortier syndrome, 619543 |
| OLA1 | 100% | 100% | 100% | 100% | 99.7% | |
| PLOD1 | 96.5% | 96.5% | 100% | 100% | 99.4% | Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 |

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| PMEPA1 | 100% | 100% | 100% | 100% | 98.5% | |
| PRKG1 | 95.9% | 95.9% | 100% | 100% | 99.6% | Aortic aneurysm, familial thoracic 8, 615436 |
| ROBO4 | 100% | 100% | 100% | 100% | 99.3% | Aortic valve disease 3, 618496 |
| SKI | 100% | 100% | 100% | 99.9% | 98.9% | Shprintzen-Goldberg syndrome, 182212 |
| SLC2A10 | 94.7% | 94.7% | 100% | 100% | 99.6% | Arterial tortuosity syndrome, 208050 |
| SMAD2 | 86.7% | 86.7% | 100% | 100% | 99.7% | Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657 |
| SMAD3 | 100% | 100% | 100% | 99.9% | 99.4% | Loeys-Dietz syndrome 3, 613795 |
| SMAD4 | 95.4% | 95.4% | 100% | 100% | 99.9% | Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 |
| SMAD6 | 100% | 100% | 100% | 100% | 98.6% | Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439 |
| TBX20 | 100% | 100% | 100% | 100% | 99.8% | Atrial septal defect 4, 611363 |
| TES | 100% | 100% | 100% | 100% | 99.4% | |
| TGFB2 | 100% | 100% | 100% | 100% | 99.8% | Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631 |

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| TGFB3 | 88.3% | 88.3% | 100% | 100% | 99.7% | Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582 |
| TGFBR1 | 100% | 100% | 100% | 100% | 99.6% | {Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192 |
| TGFBR2 | 100% | 100% | 100% | 100% | 99.5% | Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239 |
| THBS2 | 100% | 100% | 100% | 100% | 99.4% | ?Ehlers-Danlos syndrome, classic-like, 3, 620865;{Lumbar disc herniation, susceptibility to}, 603932 |
| THSD1 | 100% | 100% | 100% | 100% | 99.7% | ?Aneurysm, intracranial berry, 12, 618734;Lymphatic malformation 13, 620244 |
| THSD4 | 100% | 100% | 100% | 100% | 99.7% | Aortic aneurysm, familial thoracic 12, 619825 |

*Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*

*TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.*
*TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.*
*srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.*
*srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.*
*srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.*
*non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*
*OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.*
*This list is accurate for panel version DG 5.0.0*
*Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*